215 The Family History

CHARLES E. DRISCOLL

Definition

A family history consists of the collection of information about the patient and other family members devoted to an understanding of:

- Heritable illness
- Current family health status
- Psychosocial disorders
- Interactional and relationship data

Technique

Eliciting the family history involves questioning about several areas of a patient's life: medical, psychologic, and social. Family members or significant others may need to be considered as additional sources of information. Data collection can be most efficiently and completely accomplished using the combination of a self-administered health history questionnaire (see Figure 215.1) and a personal interview. According to Jolly and associates, the time to complete and record a thorough interview will average 16 minutes (range 9 to 30 minutes).

Timing of the family history within the context of the medical history is variable. Some elements of the family history seem most appropriate when they can be related to the present illness, such as with a presenting complaint suggesting colitis or anginal chest pain. Patients can understand the relevance of such questions if the illness under consideration has occurred in other family members. When no logical connection to the current problem can be made, inserting the family history just before or after the review of systems is most efficient and may have the effect of shortening the list of questions that need to be asked later. Opening this line of questioning can be accomplished with a bridging statement such as, "Sometimes the health of a person's family members may affect one's own health. Please tell me about the health and illnesses of your family." After responses to the open-ended question have been given, other information is gathered by direct questioning ("Has anyone in your family had a birth defect?") or using a "laundry list" question ("Has anyone in your family had an emotional illness such as anxiety, mania, depression, or schizophrenia?").

The content of the family history may be gathered in clusters of questions (see Table 215.1). The first cluster should enable the physician to understand the family composition of the patient (proband); at least three generations must be included. Names, sex, relationship to the patient, and dates (marriages, births, and deaths) are recorded for all first-degree relatives. First-degree relatives would include spouse or significant other, children, siblings, and parents. In some cases (e.g., when considering an X-linked recessive trait), information should also be gathered for more distant relatives, such as half-siblings, grandparents, uncles, and aunts. Racial and ethnic origins may give clues for the detection of genetic diseases.

FAMILY HISTORY:	If Living		If Deceased		Has any blood relative ever had:	Please Encircle No or Yes	
	Age	Health	Age at Death	Cause			Who
Father					Allergies	No Yes	
Mother					Asthma	No Yes	
Brother or Sister					Arthritis	No Yes	
1.					Glaucoma	No Yes	
2.					Cancer	No Yes	
3.					Tuberculosis	No Yes	
4.					Diabetes	No Yes	
5.					Heart Trouble	No Yes	
6.					High Blood Pressure	No Yes	
Husband or Wife					Stroke	No Yes	
Son or Daughter					Epilepsy	No Yes	
1.					Substance Abuse	No Yes	
2.					Depression	No Yes	
3.					Emotional Problems	No Yes	
4.					Suicide	No Yes	
5.					Kidney Trouble	No Yes	
6.					Thyroid Disease	No Yes	

Figure 215.1
Questions for a self-administered family history. (Reproduced with permission from Health History Questionnaire, Department of Family Practice, The University of Iowa College of Medicine.)

Table 215.1
Content Areas of Family History

Composition of family (at least 3 generations for siblings, children, parents)
Names, sex, and age or date of birth
Dates of marriages, divorces, and deaths
Racial and ethnic origins
Members of extended family
Significant others (homosexual or heterosexual partners)
Familial: heritable diseases
Genetic disorders and congenital malformations
Biochemical or metabolic disorders
Renal and cardiovascular disorders
Family psychiatric history
Neurologic disorders
Interactions, relationships, and psychosocial problems
Roles of family members
Types and strength of relationships
Major stressors
Members living together in the household

The second cluster of questions should focus on a history of familial, heritable illnesses in a family. A hereditary condition is most likely if any family member has had a condition similar to the patient's. Areas of concern are the following: deaths of infants or children, congenital malformations, biochemical or metabolic disorders, mental retardation, spontaneous abortions or infertility, unexplained or sudden deaths, any family members undergoing genetic counseling or evaluation, neurologic disorders, and a family psychiatric history. With regard to specific disorders for inquiry, supplement the list in Figure 215.1 with anemia, bleeding disorders, gout, migraine, and obesity.

A final area for investigation is the kinds of interactions and relationships among family members. Psychosocial problems should also be sought at this time. Leaving this line of questioning until the end allows for the strengthening of trust and rapport within the doctor–patient relationship necessary for successful interrogation about these potentially sensitive areas. Major stressors in the family may be responsible for physical conditions. Stomach pain or headaches in an adolescent often can stem from extreme anxiety about an alcoholic parent or impending divorce of the parents. Identify the following: Who provides the nurturing role for the family? Who is the breadwinner? Who makes decisions? Where are the conflicting, overclose (enmeshed), and domineering relationships? Who are the persons living in the immediate household?

Recording data from a family history may be best accomplished with a combination of diagrammatic and narrative forms. When genetic and interactional data are combined, the display is referred to as a *genogram*. This represents an important modification of the pedigree diagram, in which only medical diagnoses are presented. Retrieval of information about all members of a family is accomplished at a glance and may compensate for the inability to store all charts of a family together. Although a key is always included, the use of standardized symbols and notation presents the clearest message. Three or more generations are recorded with a single generation on the same horizontal line. Each generation may be designated by a roman numeral. First-born members are farthest to the left. All symbols except those representing abortions should be the same size. The male symbol is a square and traditionally is placed to the left of the round female spouse symbol. An arrow pointing to his or her symbol identifies the patient. A parent–child connection is represented with a vertical line above the symbol. A vertical line below the symbol connected by a horizontal line constitutes the marriage line. A dotted line may be used to surround all members of a family living together in the same household; interactional–relationship lines are used to denote extraordinary connections. Figure 215.2 shows the standard symbols and a typical genogram for a young asthmatic male.

Finally, it should be emphasized that a statement in the medical record such as "refer to old charts for family history" is never acceptable. Family history and relationships are dynamic and ever changing. Old charts may be a useful source of information, but updating must be done so as to detect genetic diseases or status changes that have late onset and are only acquired as aging occurs (e.g., Huntington's chorea).

Basic Science

A positive family history may clearly show an autosomal dominant, autosomal recessive, or sex-linked pattern of inheritance. In contrast, a genetically determined disorder may exist with a negative family history. The various modes of inheritance may be classified as: (1) single-gene disorders; (2) multifactorial disorders; (3) and chromosomal aberrations (see Table 215.2).

The single-gene disorders refer to autosomal dominant, autosomal recessive, sex-linked dominant, and sex-linked recessive. Nearly all sex-linked conditions are X-linked. There are more than 3000 traits known or suspected to be inherited as Mendelian or single-gene disorders (see Gelehrter, 1983). Autosomal dominant traits appear in each generation and are transmitted to approximately 50% of both male and female offspring. Homozygotes generally do not survive. Autosomal recessive traits are usually found only in siblings and not in parents or offspring. Transmission affects roughly 25% of both the male and female siblings. Consanguinity is a factor in some autosomal recessive inheritance.

The female to male ratio of sex-linked dominant disorders is 2:1. Affected males will transmit the disorder to all daughters and no sons. Heterozygous females transmit the disorder to about 50% of their children, while homozygotes transmit to all their children. With sex-linked recessive traits, males manifest the disorder more commonly than females. An affected male transmits the disorder to 50% of his daughters only after mating with a carrier female. An affected male mating with a normal female will never transmit the disease to a son, but 50% of his daughters will be carriers. The factors of nonpenetrance, variable expressivity, spontaneous mutation, and extramarital parentage may alter the expression of a genetic trait.

Multifactorial disorders are conditions that are produced by a combination of genetic and environmental factors; consequently, diagnosis is difficult. Environmental teratogens such as alcohol and drugs have been identified as two of the co-etiologic agents in some of these disorders. Much of adult genetic disease is multifactorial in origin and encompasses such health problems as hypertension, coronary artery disease, family cancer syndromes, diabetes, and epilepsy. Careful family population studies are necessary to spark the suspicion of a genetic diagnosis.

Chromosomal aberrations are of two basic types: numerical, in which extra or missing chromosomes occur; and structural, in which segmental deletion or duplication, inversion, translocation, or isochromosomes occur. Structural

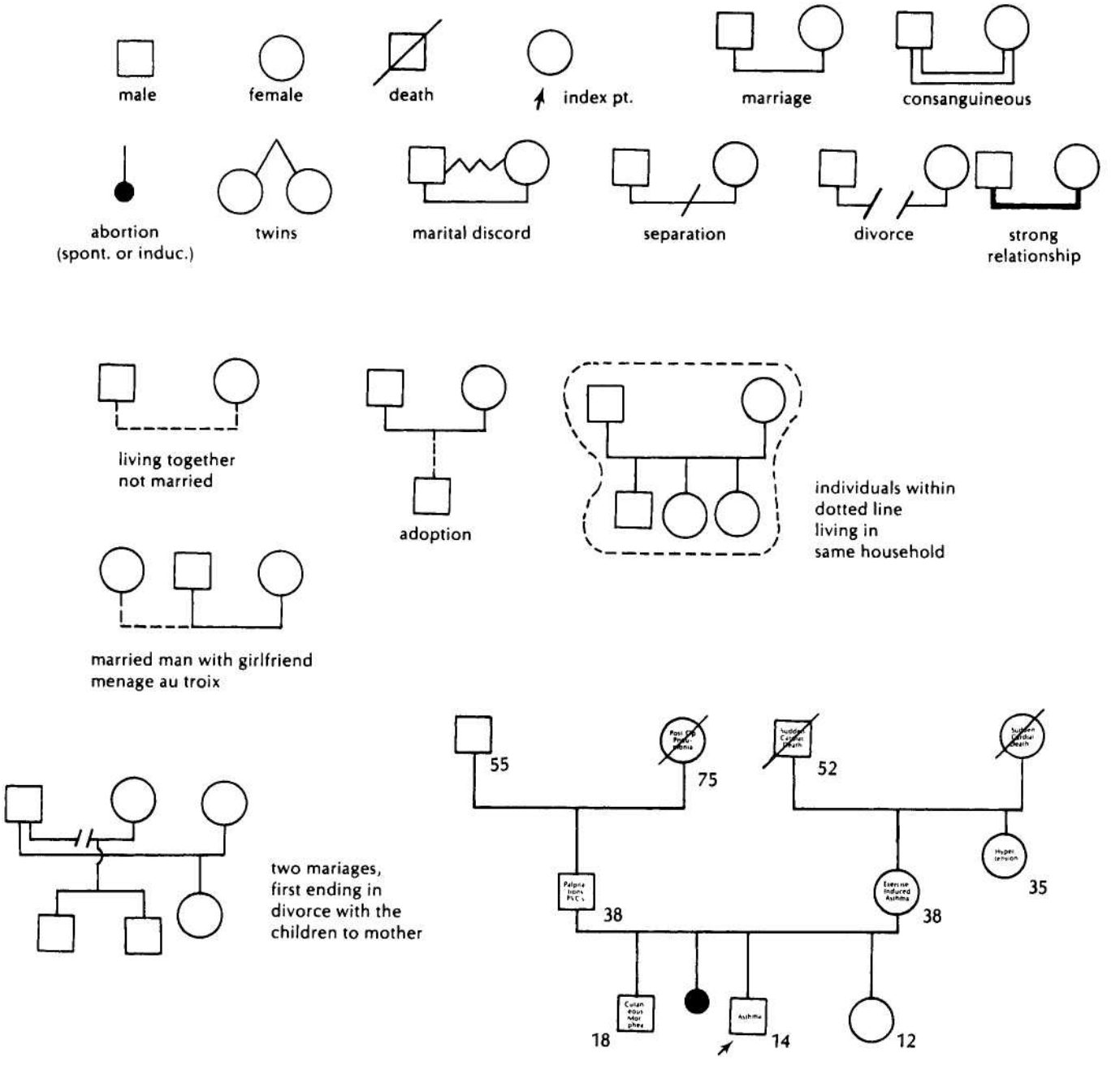

Figure 215.2
Standard symbols and notation for recording a genogram. (Reproduced with permission from Driscoll, CE, Bope E, Smith D, Carter B. The handbook of family practice. Chicago, Year Book Medical Publishers, 1986;11.)

alterations are less common than numerical ones. Numerical alterations primarily arise through the process of nondysjunction in meiosis. Nondysjunction in mitosis, after fertilization has occurred, results in two distinct cell types, referred to as *mosaicism*. Direct assessment of chromosomes by a karyotype is required for diagnosis.

Clinical Significance

Usefulness of the family history begins with the search for genetic diseases. Some genetic diseases are rare, though altogether they represent a major cause of morbidity and and mortality. It has been estimated that between 20 and 30% of pediatric hospital admissions are for some genetically related condition. For adults, this figure approaches 10%. Genetic counseling can be provided to explain the progression, prognosis, and management of current illnesses; to understand risks for recurrence and alternatives available to prevent recurrence; and to minimize the burden on the family and community of genetic diseases in a way most compatible with the patient's personal values and religious beliefs. Certain serious risks to life can be completely avoided, as in the case of cholinesterase deficiency and anesthetic accidents with the use of suxamethonium. A careful family history may provide not only help in the management

Table 215.2
Recognizable Genetic Conditions

Mendelian (single-gene) disorders
Autosomal dominant
- Achondroplasia
- Marfan's syndrome
- Retinoblastoma
- Neurofibromatosis
- Huntington's chorea
- Polycystic kidney disease (adult type)

Autosomal recessive
- Albinism
- Cystic fibrosis
- Galactosemia
- Phenylketonuria
- Sickle cell anemia
- Thalassemia

Sex-linked dominant
- Hypophosphatemia
- Amelogenesis imperfecta
- Goltz's syndrome
- Bipolar depressive psychosis
- Oral-facial-digital syndrome I

Sex-linked recessive
- Color blindness
- Duchenne's muscular dystrophy
- G_6PD deficiency
- Hemophilia A and B
- Fabry's disease

Multifactorial disorders
Asthma
Diabetes mellitus
Clubfoot
Anencephaly, spina bifida
Congenital hip dislocation
Pyloric stenosis
Cleft lip/palate
Schizophrenia

Chromosomal aberrations
Numerical
- Down's syndrome (94%)
- Klinefelter's syndrome (85%)
- Turner's syndrome (90%)

Structural
- Down's syndrome (3%)
- Cri du chat syndrome
- Wolf-Hirschhorn syndrome

Mosaicism
- Down's syndrome (3%)
- Klinefelter's syndrome (15%)
- Turner's syndrome (10%)

of a patient's disorder but also the clues for screening and identifying other at-risk patients for whom either prevention or early intervention is appropriate.

Obtaining a family psychiatric history has definite clinical benefit. A child of a schizophrenic mother has an eightfold increase in the risk for that disorder, as well as an increased risk for suicide. In the general population, anxiety neurosis has a prevalence of about 5%, which increases to about 50% in the offspring of persons with the disorder. Of persons with mania or depression, roughly two-thirds of the offspring between 5 and 15 years will be clinically depressed. In these cases, a family history succeeds in identifying unsuspected cases of illness that need treatment and may suggest to the physician that a shattered family system exists.

Finally, the detection of family roles and functioning will give assistance in achieving therapeutic goals and compliance. Not only may an individual's health or illness be a result of what happens in a family, but the family may be predictably altered by the illness of one of its members. Using family systems theory, some effective interventions may now be directed at altering the family's impact on illness. The family strengths and weaknesses, and its ability to withstand major stresses in the future, can be assessed. A good family history may reveal unexplained symptoms to be the manifestations of a stress-related or psychosomatic illness.

References

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